

ABSTRACT

The present invention relates to a novel human ABCC12 gene as well as the cDNAs encoding the novel short and long of ABCC12 proteins isoforms. The invention also relates to vectors and recombinant host cells comprising such nucleic acids, nucleotide probes and primers, and means for the detection of polymorphisms and mutations in the ABCC12 gene or in the corresponding proteins isoforms produced by the allelic form of the ABCC12 gene.